AP4 deficiency: A novel form of neurodegeneration with brain iron accumulation?

Titre	AP4 deficiency: A novel form of neurodegeneration with brain iron accumulation?
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Auteur	Roubertie, Agathe [1], Hieu, Nelson [2], Roux, Charles-Joris [3], Leboucq, Nicolas [4], Manes, Gael [5], Charif, Majida [6], Echenne, Bernard [7], Goizet, Cyril [8], Guissart, Claire [9], Meyer, Pierre [10], Marelli, Cecilia [11], Rivier, François [12], Burglen, Lydie [13], Horvath, Rita [14], Hamel, Christian [15], Lenaers, Guy [16]
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Mots-clés	AP4M1 neuropathy with barin iron accumulation [17]
Résumé en anglais	Objective: To describe the clinico-radiological phenotype of 3 patients harboring a homozygous novel pathogenic mutation. Methods: The 3 patients from an inbred family who exhibited early-onset developmental delay, tetraparesis, juvenile motor function deterioration, and intellectual deficiency were investigated by magnetic brain imaging using T1-weighted, T2-weighted, T2*-weighted, fluid-attenuated inversion recovery, susceptibility weighted imaging (SWI) sequences. Whole-exome sequencing was performed on the 3 patients. Results: In the 3 patients, brain imaging identified the same pattern of bilateral SWI hyposignal of the globus pallidus, concordant with iron accumulation. A novel homozygous nonsense mutation was identified in , segregating with the disease and leading to truncation of half of the domain of the protein. Conclusions: Our results suggest that represents a new candidate gene that should be considered in the neurodegeneration with brain iron accumulation (NBIA) spectrum of disorders and highlight the intersections between hereditary spastic paraplegia and NBIA clinical presentations.
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[1] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=16870
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